Alan H Beggs

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1829471/publications.pdf

Version: 2024-02-01

267	24,222	7096 78	9103 144
papers	citations	h-index	g-index

281	281	281	23739
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Effects of participation in a U.S. trial of newborn genomic sequencing on parents at risk for depression. Journal of Genetic Counseling, 2022, 31, 218-229.	1.6	5
2	Optical genome mapping identifies rare structural variations as predisposition factors associated with severe COVID-19. IScience, 2022, 25, 103760.	4.1	15
3	Parental Attitudes Toward Standard Newborn Screening and Newborn Genomic Sequencing: Findings From the BabySeq Study. Frontiers in Genetics, 2022, 13, 867371.	2.3	19
4	INCEPTUS Natural History, Run-in Study for Gene Replacement Clinical Trial in X-Linked Myotubular Myopathy. Journal of Neuromuscular Diseases, 2022, 9, 503-516.	2.6	5
5	X-linked myotubular myopathy is associated with epigenetic alterations and is ameliorated by HDAC inhibition. Acta Neuropathologica, 2022, 144, 537-563.	7.7	8
6	Family genetic result communication in rare and undiagnosed disease communities: Understanding the practice. Journal of Genetic Counseling, 2021, 30, 439-447.	1.6	4
7	A Cross-Sectional Study of Nemaline Myopathy. Neurology, 2021, 96, e1425-e1436.	1.1	21
8	RCL1 copy number variants are associated with a range of neuropsychiatric phenotypes. Molecular Psychiatry, 2021, 26, 1706-1718.	7.9	10
9	Underrepresentation of Phenotypic Variability of 16p13.11 Microduplication Syndrome Assessed With an Online Self-Phenotyping Tool (Phenotypr): Cohort Study. Journal of Medical Internet Research, 2021, 23, e21023.	4.3	4
10	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	2.4	34
11	Discordant results between conventional newborn screening and genomic sequencing in the BabySeq Project. Genetics in Medicine, 2021, 23, 1372-1375.	2.4	47
12	Costs and health resource use in patients with X-linked myotubular myopathy: insights from U.S. commercial claims. Journal of Managed Care & Specialty Pharmacy, 2021, 27, 1-8.	0.9	0
13	Acute and chronic tirasemtiv treatment improves in vivo and in vitro muscle performance in actin-based nemaline myopathy mice. Human Molecular Genetics, 2021, 30, 1305-1320.	2.9	11
14	Progressive cerebellar atrophy in a patient with complex II and III deficiency and a novel deleterious variant in SDHA: A Counseling Conundrum. Molecular Genetics & Genomic Medicine, 2021, 9, e1692.	1.2	1
15	Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. Molecular Genetics & Genomic Medicine, 2021, 9, e1665.	1.2	11
16	A data-driven architecture using natural language processing to improve phenotyping efficiency and accelerate genetic diagnoses of rare disorders. Human Genetics and Genomics Advances, 2021, 2, 100035.	1.7	4
17	Estimation of the Quality-of-Life Impact of X-Linked Myotubular Myopathy. Journal of Neuromuscular Diseases, 2021, 8, 1047-1061.	2.6	4
18	Psychosocial Effect of Newborn Genomic Sequencing on Families in the BabySeq Project. JAMA Pediatrics, 2021, 175, 1132.	6.2	35

#	Article	IF	Citations
19	Directed evolution of a family of AAV capsid variants enabling potent muscle-directed gene delivery across species. Cell, 2021, 184, 4919-4938.e22.	28.9	193
20	Sarcomeres regulate murine cardiomyocyte maturation through MRTF-SRF signaling. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	38
21	Artificial intelligence enables comprehensive genome interpretation and nomination of candidate diagnoses for rare genetic diseases. Genome Medicine, 2021, 13, 153.	8.2	53
22	Mortality and respiratory support in X-linked myotubular myopathy: a RECENSUS retrospective analysis. Archives of Disease in Childhood, 2020, 105, 332-338.	1.9	24
23	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
24	The Genomics Research and Innovation Network: creating an interoperable, federated, genomics learning system. Genetics in Medicine, 2020, 22, 371-380.	2.4	30
25	Prospective, phenotype-driven selection of critically ill neonates for rapid exome sequencing is associated with high diagnostic yield. Genetics in Medicine, 2020, 22, 736-744.	2.4	83
26	ASCâ€1 Is a Cell Cycle Regulator Associated with Severe and Mild Forms of Myopathy. Annals of Neurology, 2020, 87, 217-232.	5.3	12
27	Selenoprotein Nâ€related myopathy: a retrospective natural history study to guide clinical trials. Annals of Clinical and Translational Neurology, 2020, 7, 2288-2296.	3.7	18
28	Childrenâ€™s rare disease cohorts: an integrative research and clinical genomics initiative. Npj Genomic Medicine, 2020, 5, 29.	3.8	38
29	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	7.6	21
30	DYRK1A pathogenic variants in two patients with syndromic intellectual disability and a review of the literature. Molecular Genetics & Genomic Medicine, 2020, 8, e1544.	1.2	8
31	AMELIE speeds Mendelian diagnosis by matching patient phenotype and genotype to primary literature. Science Translational Medicine, 2020, 12, .	12.4	60
32	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. American Journal of Human Genetics, 2020, 106, 570-583.	6.2	37
33	Knockin mouse model of the human CFL2 p.A35T mutation results in a unique splicing defect and severe myopathy phenotype. Human Molecular Genetics, 2020, 29, 1996-2003.	2.9	5
34	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in <scp>PIGQ</scp>: Report of seven new subjects and review of the literature. Journal of Inherited Metabolic Disease, 2020, 43, 1321-1332.	3.6	15
35	Novel Recessive TNNT1 Congenital Coreâ€Rod Myopathy in French Canadians. Annals of Neurology, 2020, 87, 568-583.	5.3	19
36	Quantifying Downstream Healthcare Utilization in Studies of Genomic Testing. Value in Health, 2020, 23, 559-565.	0.3	6

#	Article	IF	Citations
37	KBTBD13 is an actin-binding protein that modulates muscle kinetics. Journal of Clinical Investigation, 2020, 130, 754-767.	8.2	25
38	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	2.4	60
39	Expanding the phenotypic spectrum associated with OPHN1 variants. European Journal of Medical Genetics, 2019, 62, 137-143.	1.3	8
40	Discovery of Novel Therapeutics for Muscular Dystrophies using Zebrafish Phenotypic Screens. Journal of Neuromuscular Diseases, 2019, 6, 271-287.	2.6	21
41	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	6.2	43
42	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clinical Imaging, 2019, 58, 108-113.	1.5	6
43	MYL2-associated congenital fiber-type disproportion and cardiomyopathy with variants in additional neuromuscular disease genes; the dilemma of panel testing. Journal of Physical Education and Sports Management, 2019, 5, a004184.	1.2	5
44	Patient-Customized Oligonucleotide Therapy for a Rare Genetic Disease. New England Journal of Medicine, 2019, 381, 1644-1652.	27.0	481
45	Returning a Genomic Result for an Adult-Onset Condition to the Parents of a Newborn: Insights From the BabySeq Project. Pediatrics, 2019, 143, S37-S43.	2.1	45
46	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. American Journal of Human Genetics, 2019, 105, 854-868.	6.2	29
47	Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. American Journal of Human Genetics, 2019, 104, 1127-1138.	6.2	59
48	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	2.5	19
49	Withdrawn Article. Molecular Genetics and Metabolism Reports, 2019, 18, 47.	1.1	0
50	IgG4â€related disease: Association with a rare gene variant expressed in cytotoxic T cells. Molecular Genetics & Genomic Medicine, 2019, 7, e686.	1.2	8
51	Unique bioinformatic approach and comprehensive reanalysis improve diagnostic yield of clinical exomes. European Journal of Human Genetics, 2019, 27, 1398-1405.	2.8	60
52	The Pediatric Cell Atlas: Defining the Growth Phase of Human Development at Single-Cell Resolution. Developmental Cell, 2019, 49, 10-29.	7.0	57
53	Aberrant regulation of epigenetic modifiers contributes to the pathogenesis in patients with selenoprotein N â€ related myopathies. Human Mutation, 2019, 40, 962-974.	2.5	13
54	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	6.2	27

#	Article	IF	Citations
55	Challenging the Current Recommendations for Carrier Testing in Children. Pediatrics, 2019, 143, S27-S32.	2.1	13
56	Perceived Benefits, Risks, and Utility of Newborn Genomic Sequencing in the BabySeq Project. Pediatrics, 2019, 143, S6-S13.	2.1	47
57	FDA oversight of NSIGHT genomic research: the need for an integrated systems approach to regulation. Npj Genomic Medicine, 2019, 4, 32.	3.8	6
58	Parental interest in genomic sequencing of newborns: enrollment experience from the BabySeq Project. Genetics in Medicine, 2019, 21, 622-630.	2.4	61
59	Interpretation of Genomic Sequencing Results in Healthy and Ill Newborns: Results from the BabySeq Project. American Journal of Human Genetics, 2019, 104, 76-93.	6.2	176
60	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	6.2	59
61	ClinPhen extracts and prioritizes patient phenotypes directly from medical records to expedite genetic disease diagnosis. Genetics in Medicine, 2019, 21, 1585-1593.	2.4	67
62	Rpl5-Inducible Mouse Model for Studying Diamond-Blackfan Anemia. Discoveries, 2019, 7, e96.	2.3	5
63	SPEG-deficient skeletal muscles exhibit abnormal triad and defective calcium handling. Human Molecular Genetics, 2018, 27, 1608-1617.	2.9	22
64	Sarcomeric and nonmuscle Î±â€actinin isoforms exhibit differential dynamics at skeletal muscle <scp>Z</scp>â€lines. Cytoskeleton, 2018, 75, 213-228.	2.0	11
65	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	6.2	59
66	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
67	Dysfunctional sarcomere contractility contributes to muscle weakness in ACTA1â€related nemaline myopathy (NEM3). Annals of Neurology, 2018, 83, 269-282.	5.3	24
68	A multicenter, retrospective medical record review of Xâ€linked myotubular myopathy: The recensus study. Muscle and Nerve, 2018, 57, 550-560.	2.2	54
69	CONGENITAL MYOPATHIES: NEMALINE AND TITINOPATHIES. Neuromuscular Disorders, 2018, 28, S105.	0.6	0
70	Novel variants in SPTAN1 without epilepsy: An expansion of the phenotype. American Journal of Medical Genetics, Part A, 2018, 176, 2768-2776.	1.2	19
71	The Genetic Landscape of Diamond-Blackfan Anemia. American Journal of Human Genetics, 2018, 103, 930-947.	6.2	184
72	De novo variant of TRRAP in a patient with very early onset psychosis in the context of non-verbal learning disability and obsessive-compulsive disorder: a case report. BMC Medical Genetics, 2018, 19, 197.	2.1	7

#	Article	IF	Citations
73	An open source microcontroller based flume for evaluating swimming performance of larval, juvenile, and adult zebrafish. PLoS ONE, 2018, 13, e0199712.	2.5	13
74	De novo ATP1A3 and compound heterozygous NLRP3 mutations in a child with autism spectrum disorder, episodic fatigue and somnolence, and muckle-wells syndrome. Molecular Genetics and Metabolism Reports, 2018, 16, 23-29.	1.1	12
75	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
76	The BabySeq project: implementing genomic sequencing in newborns. BMC Pediatrics, 2018, 18, 225.	1.7	115
77	Reconciling newborn screening and a novel splice variant in BTD associated with partial biotinidase deficiency: a BabySeq Project case report. Journal of Physical Education and Sports Management, 2018, 4, a002873.	1.2	7
78	RNA helicase, DDX27 regulates skeletal muscle growth and regeneration by modulation of translational processes. PLoS Genetics, 2018, 14, e1007226.	3.5	34
79	Newborn Sequencing in Genomic Medicine and Public Health. Pediatrics, 2017, 139, .	2.1	174
80	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	6.2	142
81	A curated gene list for reporting results of newborn genomic sequencing. Genetics in Medicine, 2017, 19, 809-818.	2.4	79
82	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	6.2	35
83	Systemic AAV8-Mediated Gene Therapy Drives Whole-Body Correction of Myotubular Myopathy in Dogs. Molecular Therapy, 2017, 25, 839-854.	8.2	81
84	Drug discovery for Diamond-Blackfan anemia using reprogrammed hematopoietic progenitors. Science Translational Medicine, 2017, 9, .	12.4	87
85	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, .	12.4	516
86	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. American Journal of Human Genetics, 2017, 100, 843-853.	6.2	181
87	Longâ€term effects of systemic gene therapy in a canine model of myotubular myopathy. Muscle and Nerve, 2017, 56, 943-953.	2.2	50
88	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	6.2	96
89	A Novel Missense Variant in the AGRN Gene; Congenital Myasthenic Syndrome Presenting With Head Drop. Journal of Clinical Neuromuscular Disease, 2017, 18, 147-151.	0.7	26
90	Novel mutation in CNTNAP1 results in congenital hypomyelinating neuropathy. Muscle and Nerve, 2017, 55, 761-765.	2.2	15

#	Article	IF	Citations
91	Homozygous EEF1A2 mutation causes dilated cardiomyopathy, failure to thrive, global developmental delay, epilepsy and early death. Human Molecular Genetics, 2017, 26, 3545-3552.	2.9	27
92	A natural history study of X-linked myotubular myopathy. Neurology, 2017, 89, 1355-1364.	1.1	75
93	Development of Soft Tissue Sarcomas in Ribosomal Proteins L5 and S24 Heterozygous Mice. Journal of Cancer, 2016, 7, 32-36.	2.5	22
94	Overlapping 16p13.11 deletion and gain of copies variations associated with childhood onset psychosis include genes with mechanistic implications for autism associated pathways: Two case reports. American Journal of Medical Genetics, Part A, 2016, 170, 1165-1173.	1.2	16
95	Muscle dysfunction in a zebrafish model of Duchenne muscular dystrophy. Physiological Genomics, 2016, 48, 850-860.	2.3	29
96	SLC6A1 Mutation and Ketogenic Diet in Epilepsy With Myoclonic-Atonic Seizures. Pediatric Neurology, 2016, 64, 77-79.	2.1	44
97	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 991-999.	6.2	68
98	A novel de novo mutation inATP1A3and childhood-onset schizophrenia. Journal of Physical Education and Sports Management, 2016, 2, a001008.	1.2	46
99	Clinical heterogeneity associated with KCNA1 mutations include cataplexy and nonataxic presentations. Neurogenetics, 2016, 17, 11-16.	1.4	26
100	Treatment with ActRIIB-mFc Produces Myofiber Growth and Improves Lifespan in the Acta1 H40Y Murine Model of Nemaline Myopathy. American Journal of Pathology, 2016, 186, 1568-1581.	3.8	23
101	Mutationâ€specific effects on thin filament length in thin filament myopathy. Annals of Neurology, 2016, 79, 959-969.	5.3	54
102	Skeletal Muscle Pathology in X-Linked Myotubular Myopathy: Review With Cross-Species Comparisons. Journal of Neuropathology and Experimental Neurology, 2016, 75, 102-110.	1.7	59
103	Gene Discovery in Congenital Myopathy. Pancreatic Islet Biology, 2016, , 39-83.	0.3	0
104	Expectation versus Reality: The Impact of Utility on Emotional Outcomes after Returning Individualized Genetic Research Results in Pediatric Rare Disease Research, a Qualitative Interview Study. PLoS ONE, 2016, 11, e0153597.	2.5	23
105	Whole Exome Sequencing Reveals DYSF, FKTN, and ISPD Mutations in Congenital Muscular Dystrophy Without Brain or Eye Involvement. Journal of Neuromuscular Diseases, 2015, 2, 87-92.	2.6	13
106	Skeletal Muscle MicroRNA and Messenger RNA Profiling in Cofilin-2 Deficient Mice Reveals Cell Cycle Dysregulation Hindering Muscle Regeneration. PLoS ONE, 2015, 10, e0123829.	2.5	9
107	Effect of levosimendan on the contractility of muscle fibers from nemaline myopathy patients with mutations in the nebulin gene. Skeletal Muscle, 2015, 5, 12.	4.2	21
108	Whole Exome Sequencing IdentifiesRAI1Mutation in a Morbidly Obese Child Diagnosed With ROHHAD Syndrome. Journal of Clinical Endocrinology and Metabolism, 2015, 100, 1723-1730.	3.6	33

#	Article	IF	Citations
109	X-linked myotubular myopathy in Rottweiler dogs is caused by a missense mutation in Exon 11 of the MTM1 gene. Skeletal Muscle, 2015, 5, 1.	4.2	46
110	Congenital and Other Structural Myopathies. , 2015, , 499-537.		3
111	Association of a NovelACTA1Mutation With a Dominant Progressive Scapuloperoneal Myopathy in an Extended Family. JAMA Neurology, 2015, 72, 689.	9.0	35
112	Muscle weakness inTPM3-myopathy is due to reduced Ca²⁺-sensitivity and impaired acto-myosin cross-bridge cycling in slow fibres. Human Molecular Genetics, 2015, 24, 6278-6292.	2.9	38
113	Gene replacement rescues severe muscle pathology and prolongs survival in myotubularin-deficient mice and dogs. Annals of Translational Medicine, 2015, 3, 257.	1.7	1
114	Muscle pathology, limb strength, walking gait, respiratory function and neurological impairment establish disease progression in the p.N155K canine model of X-linked myotubular myopathy. Annals of Translational Medicine, 2015, 3, 262.	1.7	8
115	Expanding the Phenotype Associated With theNEFLMutation. JAMA Neurology, 2014, 71, 1413.	9.0	30
116	A compound heterozygous mutation in GPD1 causes hepatomegaly, steatohepatitis, and hypertriglyceridemia. European Journal of Human Genetics, 2014, 22, 1229-1232.	2.8	38
117	Ultrasound assessment of the diaphragm: Preliminary study of a canine model of Xâ€linked myotubular myopathy. Muscle and Nerve, 2014, 50, 607-609.	2.2	12
118	Mutation Update: The Spectra of Nebulin Variants and Associated Myopathies. Human Mutation, 2014, 35, 1418-1426.	2.5	107
119	Novel Mutations Widen the Phenotypic Spectrum of Slow Skeletal/Î²-Cardiac Myosin (MYH7) Distal Myopathy. Human Mutation, 2014, 35, 868-879.	2.5	79
120	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	9.6	101
121	Bridging integrator 1 (Bin1) deficiency in zebrafish results in centronuclear myopathy. Human Molecular Genetics, 2014, 23, 3566-3578.	2.9	28
122	Gene Therapy Prolongs Survival and Restores Function in Murine and Canine Models of Myotubular Myopathy. Science Translational Medicine, 2014, 6, 220ra10.	12.4	141
123	Gait characteristics in a canine model of X-linked myotubular myopathy. Journal of the Neurological Sciences, 2014, 346, 221-226.	0.6	16
124	Mutation Update and Genotype-Phenotype Correlations of Novel and Previously Described Mutations inTPM2andTPM3Causing Congenital Myopathies. Human Mutation, 2014, 35, 779-790.	2.5	92
125	SPEG Interacts with Myotubularin, and Its Deficiency Causes Centronuclear Myopathy with Dilated Cardiomyopathy. American Journal of Human Genetics, 2014, 95, 218-226.	6.2	143
126	Kelch proteins: emerging roles in skeletal muscle development and diseases. Skeletal Muscle, 2014, 4, 11.	4.2	119

#	Article	IF	Citations
127	Approach to the diagnosis of congenital myopathies. Neuromuscular Disorders, 2014, 24, 97-116.	0.6	239
128	Differential Muscle Hypertrophy Is Associated with Satellite Cell Numbers and Akt Pathway Activation Following Activin Type IIB Receptor Inhibition in Mtm1 p.R69C Mice. American Journal of Pathology, 2014, 184, 1831-1842.	3.8	29
129	Altered translation of GATA1 in Diamond-Blackfan anemia. Nature Medicine, 2014, 20, 748-753.	30.7	243
130	Tissue Triage and Freezing for Models of Skeletal Muscle Disease. Journal of Visualized Experiments, 2014, , .	0.3	48
131	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. Journal of Clinical Investigation, 2014, 124, 4693-4708.	8.2	153
132	Novel deletion of RPL15 identified by array-comparative genomic hybridization in Diamondâ€“Blackfan anemia. Human Genetics, 2013, 132, 1265-1274.	3.8	97
133	Identification of KLHL41 Mutations Implicates BTB-Kelch-Mediated Ubiquitination as an Alternate Pathway to Myofibrillar Disruption in Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 1108-1117.	6.2	147
134	Mutations in KLHL40 Are a Frequent Cause of Severe Autosomal-Recessive Nemaline Myopathy. American Journal of Human Genetics, 2013, 93, 6-18.	6.2	186
135	Mutation of KCNJ8 in a patient with CantÃº syndrome with unique vascular abnormalities â€“ Support for the role of K(ATP) channels in this condition. European Journal of Medical Genetics, 2013, 56, 678-682.	1.3	79
136	Recessive truncating titin gene, TTN , mutations presenting as centronuclear myopathy. Neurology, 2013, 81, 1205-1214.	1.1	177
137	Loss of Catalytically Inactive Lipid Phosphatase Myotubularin-related Protein 12 Impairs Myotubularin Stability and Promotes Centronuclear Myopathy in Zebrafish. PLoS Genetics, 2013, 9, e1003583.	3.5	22
138	Selenoprotein N deficiency in mice is associated with abnormal lung development. FASEB Journal, 2013, 27, 1585-1599.	0.5	34
139	Enzyme replacement therapy rescues weakness and improves muscle pathology in mice with X-linked myotubular myopathy. Human Molecular Genetics, 2013, 22, 1525-1538.	2.9	71
140	Troponin activator augments muscle force in nemaline myopathy patients with nebulin mutations. Journal of Medical Genetics, 2013, 50, 383-392.	3.2	46
141	Deleting exon 55 from the nebulin gene induces severe muscle weakness in a mouse model for nemaline myopathy. Brain, 2013, 136, 1718-1731.	7.6	55
142	Analysis of Skeletal Muscle Defects in Larval Zebrafish by Birefringence and Touch-evoke Escape Response Assays. Journal of Visualized Experiments, 2013, , e50925.	0.3	56
143	Normal myofibrillar development followed by progressive sarcomeric disruption with actin accumulations in a mouse Cfl2 knockout demonstrates requirement of cofilin-2 for muscle maintenance. Human Molecular Genetics, 2012, 21, 2341-2356.	2.9	80
144	Î±â€Actininâ€2 deficiency results in sarcomeric defects in zebrafish that cannot be rescued by Î±â€actininâ€3 revealing functional differences between sarcomeric isoforms. FASEB Journal, 2012, 26, 1892-1908.	0.5	34

#	Article	IF	Citations
145	Modeling the human MTM1 p.R69C mutation in murine Mtm1 results in exon 4 skipping and a less severe myotubular myopathy phenotype. Human Molecular Genetics, 2012, 21, 811-825.	2.9	54
146	Myotubularin-Deficient Myoblasts Display Increased Apoptosis, Delayed Proliferation, and Poor Cell Engraftment. American Journal of Pathology, 2012, 181, 961-968.	3.8	37
147	Congenital myopathy caused by a novel missense mutation in the CFL2 gene. Neuromuscular Disorders, 2012, 22, 632-639.	0.6	49
148	Muscle function in A canine model of Xâ€linked myotubular myopathy. Muscle and Nerve, 2012, 46, 588-591.	2.2	23
149	Dominant Mutation of CCDC78 in a Unique Congenital Myopathy with Prominent Internal Nuclei and Atypical Cores. American Journal of Human Genetics, 2012, 91, 365-371.	6.2	84
150	Exome Sequencing and Functional Validation in Zebrafish Identify GTDC2 Mutations as a Cause of Walker-Warburg Syndrome. American Journal of Human Genetics, 2012, 91, 541-547.	6.2	167
151	Clinical utility gene card for: Centronuclear and myotubular myopathies. European Journal of Human Genetics, 2012, 20, 1101-1101.	2.8	28
152	Myotubular myopathy and the neuromuscular junction: a novel therapeutic approach from mouse models. DMM Disease Models and Mechanisms, 2012, 5, 852-9.	2.4	43
153	Mutation spectrum in the large GTPase dynamin 2, and genotype-phenotype correlation in autosomal dominant centronuclear myopathy. Human Mutation, 2012, 33, 949-959.	2.5	115
154	Frameshift mutation in p53 regulatorRPL26is associated with multiple physical abnormalities and a specific pre-ribosomal RNA processing defect in diamond-blackfan anemia. Human Mutation, 2012, 33, 1037-1044.	2.5	135
155	Mutations in the satellite cell gene MEGF10 cause a recessive congenital myopathy with minicores. Neurogenetics, 2012, 13, 115-124.	1.4	68
156	Exome sequencing identifies GATA1 mutations resulting in Diamond-Blackfan anemia. Journal of Clinical Investigation, 2012, 122, 2439-2443.	8.2	292
157	A Splice Site Mutation in Laminin-Î±2 Results in a Severe Muscular Dystrophy and Growth Abnormalities in Zebrafish. PLoS ONE, 2012, 7, e43794.	2.5	48
158	Changes in cross-bridge cycling underlie muscle weakness in patients with tropomyosin 3-based myopathy. Human Molecular Genetics, 2011, 20, 2015-2025.	2.9	60
159	Inhibition of Activin Receptor Type IIB Increases Strength and Lifespan in Myotubularin-Deficient Mice. American Journal of Pathology, 2011, 178, 784-793.	3.8	63
160	Ddx18 is essential for cell-cycle progression in zebrafish hematopoietic cells and is mutated in human AML. Blood, 2011, 118, 903-915.	1.4	43
161	Novel mutations in NEB cause abnormal nebulin expression and markedly impaired muscle force generation in severe nemaline myopathy. Skeletal Muscle, 2011, 1, 23.	4.2	51
162	The zebrafish dag1 mutant: a novel genetic model for dystroglycanopathies. Human Molecular Genetics, 2011, 20, 1712-1725.	2.9	101

#	Article	IF	Citations
163	Myotubularin controls desmin intermediate filament architecture and mitochondrial dynamics in human and mouse skeletal muscle. Journal of Clinical Investigation, 2011, 121, 70-85.	8.2	132
164	Mutations of tropomyosin 3 (TPM3) are common and associated with type 1 myofiber hypotrophy in congenital fiber type disproportion. Human Mutation, 2010, 31, 176-183.	2.5	70
165	The ribosomal basis of diamond-blackfan anemia: mutation and database update. Human Mutation, 2010, 31, 1269-1279.	2.5	202
166	Functional muscle analysis of the Tcap knockout mouse. Human Molecular Genetics, 2010, 19, 2268-2283.	2.9	45
167	MTM1 mutation associated with X-linked myotubular myopathy in Labrador Retrievers. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 14697-14702.	7.1	114
168	Cell Membrane Expression of Cardiac Sodium Channel Na_v1.5 Is Modulated by Î±-Actinin-2 Interaction. Biochemistry, 2010, 49, 166-178.	2.5	57
169	Altered myofilament function depresses force generation in patients with nebulin-based nemaline myopathy (NEM2). Journal of Structural Biology, 2010, 170, 334-343.	2.8	87
170	T-tubule disorganization and defective excitation-contraction coupling in muscle fibers lacking myotubularin lipid phosphatase. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 18763-18768.	7.1	167
171	Thin filament length dysregulation contributes to muscle weakness in nemaline myopathy patients with nebulin deficiency. Human Molecular Genetics, 2009, 18, 2359-2369.	2.9	124
172	Serotonin-Related FEV Gene Variant in the Sudden Infant Death Syndrome Is a Common Polymorphism in the African-American Population. Pediatric Research, 2009, 66, 631-635.	2.3	16
173	Mutations and polymorphisms of the skeletal muscle ÃŽÂ±-actin gene (ACTA1). Human Mutation, 2009, 30, 1267-1277.	2.5	198
174	Fastâ€twitch sarcomeric and glycolytic enzyme protein loss in inclusion body myositis. Muscle and Nerve, 2009, 39, 739-753.	2.2	41
175	Automated DNA mutation detection using universal conditions direct sequencing: application to ten muscular dystrophy genes. BMC Genetics, 2009, 10, 66.	2.7	13
176	The exon 55 deletion in the nebulin gene â€“ One single founder mutation with world-wide occurrence. Neuromuscular Disorders, 2009, 19, 179-181.	0.6	54
177	Ribosomal Protein Genes S10 and S26 Are Commonly Mutated in Diamond-Blackfan Anemia.. Blood, 2009, 114, 175-175.	1.4	2
178	Ribosomal Protein L5 and L11 Mutations Are Associated with Cleft Palate and Abnormal Thumbs in Diamond-Blackfan Anemia Patients. American Journal of Human Genetics, 2008, 83, 769-780.	6.2	363
179	Erratum to â€œMyofiber size correlates with MTM1 mutation type and outcome in X-linked myotubular myopathyâ€•[Neuromuscular Disorders 17(7) (2007) 562â€“568]. Neuromuscular Disorders, 2008, 18, 519.	0.6	0
180	AAV-mediated intramuscular delivery of myotubularin corrects the myotubular myopathy phenotype in targeted murine muscle and suggests a function in plasma membrane homeostasis. Human Molecular Genetics, 2008, 17, 2132-2143.	2.9	115

#	Article	IF	Citations
181	Dynamic regulation of endothelial NOS mediated by competitive interaction with Î±â€actininâ€4 and calmodulin. FASEB Journal, 2008, 22, 1450-1457.	0.5	7
182	Acute appendicitis is characterized by a uniform and highly selective pattern of inflammatory gene expression. Mucosal Immunology, 2008, 1, 297-308.	6.0	34
183	Abnormalities of the large ribosomal subunit protein, Rpl35a, in Diamond-Blackfan anemia. Blood, 2008, 112, 1582-1592.	1.4	208
184	Remission in Patients with Diamond Blackfan Anemia (DBA) Appears to Be Unrestricted by Phenotype or Genotype. Blood, 2008, 112, 3092-3092.	1.4	3
185	Morphologic Assessment of Muscle Biopsy Abnormalities in Patients with Congenital Fiber Type Disproportion and Tropomyosin 3 Mutation. FASEB Journal, 2008, 22, 708.24.	0.5	0
186	Identification of New Rare Sequence Changes in RP Genes in Diamond-Blackfan Anemia and Association of the RPL5 and RPL11 Mutations with Craniofacial and Thumb Malformations. Blood, 2008, 112, 39-39.	1.4	0
187	Distinctive patterns of microRNA expression in primary muscular disorders. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 17016-17021.	7.1	458
188	Myofiber size correlates with MTM1 mutation type and outcome in X-linked myotubular myopathy. Neuromuscular Disorders, 2007, 17, 562-568.	0.6	39
189	Nemaline Myopathy with Minicores Caused by Mutation of the CFL2 Gene Encoding the Skeletal Muscle Actinâ€“Binding Protein, Cofilin-2. American Journal of Human Genetics, 2007, 80, 162-167.	6.2	213
190	Type I interferonâ€“inducible gene expression in blood is present and reflects disease activity in dermatomyositis and polymyositis. Arthritis and Rheumatism, 2007, 56, 3784-3792.	6.7	264
191	Mutations of the Genes for Ribosomal Proteins L5 and L11 Are a Common Cause of Diamond-Blackfan Anemia.. Blood, 2007, 110, 421-421.	1.4	8
192	Selenoproteins and Their Impact on Human Health Through Diverse Physiological Pathways. Physiology, 2006, 21, 307-315.	3.1	136
193	Ribosomal Protein S24 Gene Is Mutated in Diamond-Blackfan Anemia. American Journal of Human Genetics, 2006, 79, 1110-1118.	6.2	257
194	Multiplex PCR for Identifying DMD Gene Deletions. Current Protocols in Human Genetics, 2006, 49, Unit 9.3.	3.5	7
195	Adult-Onset Nemaline Myopathy and Monoclonal Gammopathy. Archives of Neurology, 2006, 63, 132.	4.5	39
196	Multiple Serotonergic Brainstem Abnormalities in Sudden Infant Death Syndrome. JAMA - Journal of the American Medical Association, 2006, 296, 2124.	7.4	443
197	Sporadic, Adult-Onset Nemaline Myopathy Presenting as Camptocormia (Bent-Spine Syndrome). Journal of Clinical Neuromuscular Disease, 2006, 8, 7-11.	0.7	0
198	Defective Ribosomal Protein Gene Expression Alters Transcription, Translation, Apoptosis, and Oncogenic Pathways in Diamond-Blackfan Anemia. Stem Cells, 2006, 24, 2034-2044.	3.2	75

#	Article	IF	Citations
199	Skeletal muscle repair in a mouse model of nemaline myopathy. Human Molecular Genetics, 2006, 15, 2603-2612.	2.9	44
200	Melanoma cell adhesion molecule is a novel marker for human fetal myogenic cells and affects myoblast fusion. Journal of Cell Science, 2006, 119, 3117-3127.	2.0	34
201	X-Linked Myotubular and Centronuclear Myopathies. Journal of Neuropathology and Experimental Neurology, 2005, 64, 555-564.	1.7	90
202	Mutations in dynamin 2 cause dominant centronuclear myopathy. Nature Genetics, 2005, 37, 1207-1209.	21.4	390
203	Variations in gene expression among different types of human skeletal muscle. Muscle and Nerve, 2005, 32, 483-491.	2.2	28
204	The influence of muscle type and dystrophin deficiency on murine expression profiles. Mammalian Genome, 2005, 16, 739-748.	2.2	35
205	Evidence by molecular profiling for a placental origin of infantile hemangioma. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 19097-19102.	7.1	170
206	Severe arrhythmia disorder caused by cardiac L-type calcium channel mutations. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 8089-8096.	7.1	558
207	Side Population cells isolated from different tissues share transcriptome signatures and express tissue-specific markers. Experimental Cell Research, 2005, 303, 360-374.	2.6	45
208	Defective Ribosomal Protein Gene Expression Alters Transcription, Translation and Oncogenic Pathways in Diamond-Blackfan Anemia.. Blood, 2005, 106, 3546-3546.	1.4	5
209	Transcriptional profile of postmortem skeletal muscle. Physiological Genomics, 2004, 16, 222-228.	2.3	38
210	Expression profiling and identification of novel genes involved in myogenic differentiation. FASEB Journal, 2004, 18, 1-23.	0.5	157
211	RNA and protein evidence for haplo-insufficiency in Diamond-Blackfan anaemia patients with RPS19 mutations. British Journal of Haematology, 2004, 127, 105-113.	2.5	96
212	Heterogeneity of nemaline myopathy cases with skeletal muscle Î±â€actin gene mutations. Annals of Neurology, 2004, 56, 86-96.	5.3	135
213	Genotype?phenotype correlations in nemaline myopathy caused by mutations in the genes for nebulin and skeletal muscle ?-actin. Neuromuscular Disorders, 2004, 14, 461-470.	0.6	107
214	Molecular classification of nemaline myopathies: â€œnontypingâ€•specimens exhibit unique patterns of gene expression. Neurobiology of Disease, 2004, 15, 590-600.	4.4	13
215	Gene Expression Changes in Bone Marrow Cells from Diamond-Blackfan Anemia Patients.. Blood, 2004, 104, 720-720.	1.4	8
216	Deficiency of Muscle Î±-Actinin-3 is Compatible with High Muscle Performance. Journal of Molecular Neuroscience, 2003, 20, 39-42.	2.3	9

#	Article	IF	Citations
217	Gene expression profiling of Duchenne muscular dystrophy skeletal muscle. Neurogenetics, 2003, 4, 163-171.	1.4	82
218	Reproducibility of gene expression across generations of Affymetrix microarrays. BMC Bioinformatics, 2003, 4, 27.	2.6	67
219	Filamin C accumulation is a strong but nonspecific immunohistochemical marker of core formation in muscle. Journal of the Neurological Sciences, 2003, 206, 71-78.	0.6	54
220	ACTN3 Genotype Is Associated with Human Elite Athletic Performance. American Journal of Human Genetics, 2003, 73, 627-631.	6.2	708
221	Muscle disease caused by mutations in the skeletal muscle alpha-actin gene (ACTA1). Neuromuscular Disorders, 2003, 13, 519-531.	0.6	192
222	Expression profiling reveals altered satellite cell numbers and glycolytic enzyme transcription in nemaline myopathy muscle. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 4666-4671.	7.1	68
223	Rod Distribution and Muscle Fiber Type Modification in the Progression of Nemaline Myopathy. Journal of Child Neurology, 2003, 18, 235-240.	1.4	13
224	Gene expression comparison of biopsies from Duchenne muscular dystrophy (DMD) and normal skeletal muscle. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 15000-15005.	7.1	312
225	Variant of SCN5A Sodium Channel Implicated in Risk of Cardiac Arrhythmia. Science, 2002, 297, 1333-1336.	12.6	506
226	Telethonin protein expression in neuromuscular disorders. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2002, 1588, 33-40.	3.8	49
227	Clinical and genetic heterogeneity in nemaline myopathy â€“ a disease of skeletal muscle thin filaments. Trends in Molecular Medicine, 2001, 7, 362-368.	6.7	145
228	Evidence for linkage of familial Diamond-Blackfan anemia to chromosome 8p23.3-p22 and for non-19q non-8p disease. Blood, 2001, 97, 2145-2150.	1.4	84
229	Nemaline myopathy: A clinical study of 143 cases. Annals of Neurology, 2001, 50, 312-320.	5.3	236
230	Genomic organization and single-nucleotide polymorphism map of desmuslin, a novel intermediate filament protein on chromosome 15q26.3. BMC Genetics, 2001, 2, 8.	2.7	19
231	Genotype-Phenotype Correlation in the Long-QT Syndrome. Circulation, 2001, 103, 89-95.	1.6	1,641
232	Mutations in ACTN4, encoding Î±-actinin-4, cause familial focal segmental glomerulosclerosis. Nature Genetics, 2000, 24, 251-256.	21.4	1,124
233	Mutations in the nebulin gene associated with autosomal recessive nemaline myopathy. Proceedings of the National Academy of Sciences of the United States of America, 1999, 96, 2305-2310.	7.1	304
234	Mutations in the skeletal muscle Î±-actin gene in patients with actin myopathy and nemaline myopathy. Nature Genetics, 1999, 23, 208-212.	21.4	389

#	Article	IF	Citations
235	A common nonsense mutation results in Î±-actinin-3 deficiency in the general population. Nature Genetics, 1999, 21, 353-354.	21.4	378
236	Sodium channel abnormalities are infrequent in patients with long QT Syndrome: Identification of two novelSCN5A mutations. , 1999, 86, 470-476.		48
237	Clinical and genetic heterogeneity in autosomal recessive nemaline myopathy. Neuromuscular Disorders, 1999, 9, 564-572.	0.6	84
238	Human Neuropilin-1 and Neuropilin-2 Map to 10p12 and 2q34, Respectively. Genomics, 1999, 57, 459-460.	2.9	23
239	Î±-Actinin-2 Is a New Component of the Dystrophinâ€“Glycoprotein Complex. Archives of Biochemistry and Biophysics, 1999, 365, 216-222.	3.0	40
240	Multiple different missense mutations in the pore region of HERG in patients with long QT syndrome. Human Genetics, 1998, 102, 265-272.	3.8	57
241	Congenital Fibrosis of the Extraocular Muscles Type 2, an Inherited Exotropic Strabismus Fixus, Maps to Distal 11q13. American Journal of Human Genetics, 1998, 63, 517-525.	6.2	89
242	Human Skeletal Muscle-Specific Î±-Actinin-2 and -3 Isoforms Form Homodimers and Heterodimersin Vitroandin Vivo. Biochemical and Biophysical Research Communications, 1998, 248, 134-139.	2.1	69
243	Mutation of the Gene for I sK Associated With Both Jervell and Lange-Nielsen and Romano-Ward Forms of Long-QT Syndrome. Circulation, 1998, 97, 142-146.	1.6	205
244	Molecular genetics of long-QT syndrome. Current Opinion in Pediatrics, 1998, 10, 628-634.	2.0	13
245	Differential Regional Expression and Ultrastructural Localization of Î±-Actinin-2, a Putative NMDA Receptor-Anchoring Protein, in Rat Brain. Journal of Neuroscience, 1998, 18, 1383-1392.	3.6	164
246	The Concomitant Use of Dystrophin and Utrophin/Dystrophin Related Protein Antibodies to Reduce Misdiagnosis of Duchenne/Becker Muscular Dystrophy. Biochemical and Biophysical Research Communications, 1997, 241, 232-235.	2.1	1
247	Competitive binding of Î±-actinin and calmodulin to the NMDA receptor. Nature, 1997, 385, 439-442.	27.8	567
248	Oculomotor nerve and muscle abnormalities in congenital fibrosis of the extraocular muscles. Annals of Neurology, 1997, 41, 314-325.	5.3	165
249	Dystrophinopathy, The Expanding Phenotype. Circulation, 1997, 95, 2344-2347.	1.6	47
250	Deficiency of a skeletal muscle isoform of Î±-actinin (Î±-actinin-3) in merosin-positive congenital muscular dystrophy. Neuromuscular Disorders, 1996, 6, 229-235.	0.6	127
251	The Mouse Region Syntenic for Human Spinal Muscular Atrophy Lies within theLgn1Critical Interval and Contains Multiple Copies ofNaipExon 5. Genomics, 1996, 38, 405-417.	2.9	44
252	Congenital Muscular Dystrophy Associated With Merosin Deficiency. Journal of Child Neurology, 1996, 11, 291-295.	1.4	24

#	Article	IF	Citations
253	Isoform Cloning, Actin Binding, and Chromosomal Localization of Human Erythroid Dematin, a Member of the Villin Superfamily. Journal of Biological Chemistry, 1995, 270, 17407-17413.	3.4	66
254	A severe muscular dystrophy patient with an internally deleted very short (110 kD) Dystrophin: Presence of the binding site for dystrophin-associated glycoprotein (DAG) may not be enough for physiological function of dystrophin. Neuromuscular Disorders, 1995, 5, 429-438.	0.6	14
255	Novel actin crosslinker superfamily member identified by a two step degenerate PCR procedure. FEBS Letters, 1995, 368, 500-504.	2.8	44
256	Expression of the Myelin Basic Protein Gene in Transgenic Mice Expressing Human Neurotropic Virus, JCV, Early Protein. Virology, 1994, 202, 89-96.	2.4	19
257	Mapping a gene for congenital fibrosis of the extraocular muscles to the centromeric region of chromosome 12. Nature Genetics, 1994, 7, 69-73.	21.4	104
258	Human pituitary adenomas show no loss of heterozygosity at the retinoblastoma gene locus. Journal of Clinical Endocrinology and Metabolism, 1994, 78, 922-927.	3.6	61
259	Severe nonspecific X-linked mental retardation caused by a proximally Xp located gene: Intragenic heterogeneity or a new form of X-linked mental retardation?. American Journal of Medical Genetics Part A, 1993, 46, 172-175.	2.4	24
260	Prediction of dystrophin phenotype by DNA analysis in Duchenne/Becker muscular dystrophy. Pediatric Neurology, 1992, 8, 432-436.	2.1	10
261	Early onset autosomal dominant progressive muscular dystrophy presenting in childhood as a Becker phenotypeâ€"The importance of dystrophin and molecular genetic analysis. Neuromuscular Disorders, 1992, 2, 121-124.	0.6	8
262	Additional dystrophin fragment in Becker muscular dystrophy may result from proteolytic cleavage at deletion junctions. American Journal of Medical Genetics Part A, 1992, 44, 378-381.	2.4	9
263	Preservation of the C-terminus of dystrophin molecule in the skeletal muscle from Becker muscular dystrophy. Journal of the Neurological Sciences, 1991, 101, 148-156.	0.6	91
264	A polymorphic CACA repeat in the 3â€² untranslated region of dystrophin. Nucleic Acids Research, 1990, 18, 1931-1931.	14.5	85
265	Duchenne muscular dystrophy (Oxford monographs on medical genetics, Vol. 15) (revised edn). Trends in Genetics, 1989, 5, 232.	6.7	2
266	Effect of ageing on reactivation of the human X-linked HPRT locus. Nature, 1988, 335, 93-96.	27.8	81
267	Reactivation of X-linked genes in human fibroblasts transformed by origin-defective SV40. Somatic Cell and Molecular Genetics, 1986, 12, 585-594.	0.7	15